

2nd European Otolaryngology ENT Surgery Conference & International Conference on Craniofacial Surgery

Kang-Min Ahn et al., J Otol Rhinol, 6:6
DOI: 10.4172/2324-8785-C1-005



October 16-18, 2017 Rome, Italy

Novel *PTCH 1* gene mutation in nevoid basal cell carcinoma syndrome: Results of five cases

Kang-Min Ahn, Gu-Hwan Kim and Myoung-Sang You
Seoul Asan Medical Center, South Korea

Purpose: The purpose of the study is to report clinical characteristics, surgical results, and new *PTCH1* gene mutations in nevoid basal cell carcinoma syndrome (NBCCS).

Materials & Methods: Five patients were referred to the Department of Oral and Maxillofacial Surgery from local dental clinics between 2006 and 2016 to treat multiple keratocystic odontogenic tumor (KOT). The cystic lesions were enucleated and peripheral ostectomy was performed to obtain safety margin. Recurrence and/or *de novo* development of KOT were assessed. Gene analysis using peripheral blood was performed in all patients to identify the mutation of *PTCH 1* gene.

Results: Three patients showed familial history of first degree relative. Of the major criteria, all patients presented KOT but only one patient had BCC. Of the minor criteria, four of the five patients presented macrocephaly and hypertelorism. During follow-up periods, all patients showed recurrence and/or *de novo* development of KOT in the jaw bone. Mutation analysis of *PTCH1* gene showed three frameshifts (c.817_818ins(T), c.1226_1227ins(A), c.2748del(C)), one splicing (c.1504-2A>T) and one missense (c.385T>C) mutation. Mutations were found in exon 1, 6, 9, 17, and intron 10.

Conclusion: Five new mutations of *PTCH1* gene were found in this study. Our results support previous studies that indicate the importance of high family history. Regular follow-up is necessary because recurrence rate was very high. To help early diagnosis, it is essential to routinely perform genetic testing to detect *PTCH1* gene mutations among NBCCS patients.

Biography

Kang-Min Ahn was awarded PhD in Oral and Maxillofacial Surgery from Seoul National University in the 2005. He holds a Master's Degree (MSc) in Oral and Maxillofacial Surgery from Seoul National University, Korea in 2000. Currently, he is working as a Professor for Asan Medical Center, College of Medicine, University of Ulsan. His international experience includes various programs, contributions and participation in different countries for diverse fields of study. His research interests reflect in his wide range of publications in various national and international journals. He is the Board Member of Korean Association of Cleft Lip and Palate since 1996, and serves as a member of various associations.

ahnkangmin@hanmail.net

Notes: